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The dental management of an albino adolescent with an undetected bleeding disorder: a case report

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Abstract

Albinism is an autosomal recessive genetic disorder characterised by hypopigmentation of the eyes, skin and hair. It has been linked to other conditions such as Hermansky-Pudlak Syndrome (HPS). HPS is relevant to the dental profession because of the platelet disorder associated with it. This report describes the management of a 14-year-old boy with albinism. It highlights the possible link between albinism and HPS. It emphasises how important it is that dentists are aware of the medical problems associated with HPS.

Key words: *Community Special Care/Paediatric dentistry, medically compromised, oral medicine/oral surgery, prevention*

Introduction

Albinism is an autosomal recessive genetic disorder characterised by hypopigmentation of the eyes, skin and hair (oculocutaneous albinism). Its prevalence worldwide is 1:17,000 with a male to female ratio of 1:1 (Gronskov *et al*., 2007). In albinism, the body is unable to produce or distribute melanin because of one of several possible genetic defects. In type 1 albinism, there is a genetic defect in the enzyme tyrosinase which is responsible for the body's failure to convert tyrosine into melanin. Type 2 albinism is due to a defect in the 'P' gene.

Oculocutaneous albinism is the most severe form of albinism and is inherited autosomal recessively. Hypopigmentation of the skin, eyes and hair as well as vision defects are evident. In ocular albinism the eyes are affected in isolation. This is inherited either via an x-linked or an autosomal recessive pattern of inheritance (Oetting *et al*., 1996).

Complex conditions including Hermansky-Pudlak Syndrome (HPS), Chediak-Higashi Syndrome (Introne *et al*., 1999), Griscelli Syndrome and Waardenberg Syndrome have been associated with albinism (Wiktop *et al*., 1990, Introne *et al*., 1999; Ménasché *et al*., 2002). HPS is a group of complex disorders characterised by oculocutaneous albinism, a bleeding tendency and lysosomal ceroid storage resulting from defects of multiple cytoplasmic organelles: melanosomes, lysosomes and platelet dense bodies (Huizing *et al*., 2001; Huizing and Gahl, 2002). It is inherited as an autosomal recessive trait and exists with loci heterogeneity. To date, eight genetically distinct forms of HPS have been identified in the human population (Dell'Angelica et al., 1999). It is a rare condition with a worldwide prevalence of 1:500,000. However, there are areas where HPS is more common. For example in the Puerto Rican population the prevalence is 1:1,800. It is also more common in an isolated Swiss Alps village (Schallreuter *et al*., 1993).

With the increase in emigration, this condition could be seen more frequently by health care professionals in the UK.

Case Report

A 14-year-old Albino Caucasian male was referred by his general dental practitioner with a buccal and palatal bony swelling in his upper right quadrant. His medical history revealed mild asthma, weak eyesight associated with the albinism and an allergy to penicillin. The patient was denied pinnaplasty treatment by a cosmetic surgeon because of a suspected coagulation disorder. His identical twin brother had no history of a bleeding disorder. He smoked one cigarette a day.

Dental history revealed that he had multiple primary teeth extracted under general anaesthesia at the ages of 3 and 7 years. On both occasions, the surgery and recovery were unremarkable. The patient had a history of failed dental appointments. He admitted to a frequent intake of non-milk extrinsic sugars between meals. He brushed his teeth twice daily with 1450ppm fluoride toothpaste.

On extra oral examination, oculocutaneous hypopigmentation, nystagmus and bat ears were observed. Intraoral examination revealed a bony swelling in the upper right quadrant; retained 52, 53 and 54; unerrupted 11, 12, 13, 14, 17, 27, 35, 44 and 45. Caries was evident in all four first permanent molars and 47. Radiographic examination showed that all teeth were present. A well-corticated radiolucent lesion in the upper right quadrant measuring approximately 3cm in diameter encompassed the 11, 12, 13 and 14. The eruption of 17 and 27 was impeded by the overlying 18 and 28 (*Figures 1 and 2*).

A full standard haematological examination was conducted following the concerns raised by his plastic surgeon. The coagulation factor activity and platelet counts were found to be within the normal range. There was a slightly prolonged APTT of 1.28 (normal range = 0.85 to 1.15)

The patient was enrolled on a comprehensive prevention programme. Conservative treatment of the restorable permanent teeth (16, 26 and 47) was undertaken using local anaesthesia. Under general anaesthetic, the cyst-like lesion was explored and 12, 14, 18, 36, 46, 52, 53 and 54 were extracted. During surgery, the patient experienced excessive bleeding. It was, therefore, decided that the 28 should be left in situ. Haemostasis was achieved using a haemostatic pack, sutures and tranexamic acid. Healing was normal and uneventful. Histopathological examination confirmed that the lesion was a dentigerous cyst.

Discussion

Healthcare professionals should be aware that albinism can be associated with other conditions such as HPS. HPS is a rare autosomal recessive inherited disorder. It is characterised by oculocutaneous albinism, platelet dysfunction and ceroid storage. Individuals with this condition may suffer with a bleeding disorder, cardiomyopathies, diabetes, pulmonary fibrosis and renal failure.

It has been reported that individuals with HPS may present with a range of skin, hair and eye colours. Skin may be white or creamy as found in type 1 albinism or freckled. Hair colour can vary from yellow, light brown or dark brown with blue or brown eyes. These differences from type 1 albinism could help when making an initial diagnosis of HPS in the clinical dental setting (Gronskov *et al*., 2007).

HPS is a rare disorder but dentists ought to be familiar with it nonetheless because it has serious medical implications. The bleeding disorder varies in onset and degree of severity between individuals. Onset may range from early childhood through to adulthood. Sufferers can experience mild bruising to life-threatening bleeding. One author described a case where a patient bled to death after a tooth was extracted (Theuring and Fiedler, 1973).

In HPS patients, standard haematological investigation results are normal including PT, PTT and platelet count. The bleeding-time is usually prolonged but may be normal. To confirm the diagnosis it is necessary to examine the platelets under a whole mount electron microscope for the presence of dense bodies. In HPS platelets have a reduced number or total absence of dense bodies (Witkop *et al*.,1987). When injury occurs, platelets release their dense bodies which discharge ADP, ATP, serotonin, calcium and phosphate. These components attract further platelets to the site of vessel damage to form a platelet plug. Additionally subjects with HPS have significantly lower Von Willebrand Factor (vWF) activity levels and slightly lower values of plasma vWF activity when compared to subjects without HPS (Hussain *et al*., 1998; McKeon *et al*., 1998). Any individual with albinism and a suspected bleeding disorder should be investigated by a haematologist for HPS. Early diagnosis is important in order to prevent and manage serious situations appropriately.

A comprehensive prevention programme is of primary importance in order to avoid the need for invasive dental procedures in HPS. Ideally, this should be introduced at an early age. It is essential to adopt a high quality of dental care to prevent the need for dental extractions. Consultation with the haematologist should be considered mandatory before undertaking any invasive dental treatment. Local or systemic measures such as tranexamic acid, haemostatic pack and sutures should be prepared carefully in order to reduce the risk of excessive bleeding. Aspirin and other non-steroidal anti-inflammatory drugs should be avoided and alternative medication such as paracetamol should be used instead to provide analgesia. In HPS patients, a lipid-protein ceroid complex accumulates in several organs around the body. In the mouth, mild fibrotic gingival enlargement may be present. There may also be cardiomyopathies, diabetes, inflammatory bowel disease, pulmonary fibrosis and renal failure. The accumulation of ceroid is associated with an inflammatory response in the lungs. This eventually scars the lung tissue causing it to lose its ability to expand and contract whilst breathing. Pulmonary fibrosis is the most common cause of death in patients with HPS. It tends to manifest in the thirties (Gahl *et al*., 1998; Brantly *et al*., 2000; Avila *et al*., 2002).

Future Management

The patient has been referred to the Department of Haematology to establish whether or not he has HPS. This is imperative in order to effectively plan future health management. His twin brother has been advised to undergo screening for this condition. Meticulous prevention, including encouraging the patient to enrol in a smoking cessation programme has been implemented. This is extremely important as, in the future, the patient may develop pulmonary fibrosis, one of the complications of HPS.

Conclusions

This case highlights the possible link between albinism and HPS. An individual with this condition may suffer from cardiomyopathies, diabetes, pulmonary fibrosis, renal failure and a platelet dysfunction which, crucially, is not detected using standard haematological tests. The dentist may be the first health professional to whom HPS presents. It is important that dentists are aware of the association between these conditions and their medical implications so that appropriate precautions can be taken to ensure that the patient is managed safely.

Figure 1. A dentopantomograph (DPT) radiograph showing a well-corticated radiolucent lesion encompassing the displaced 11,12,13 and 14.

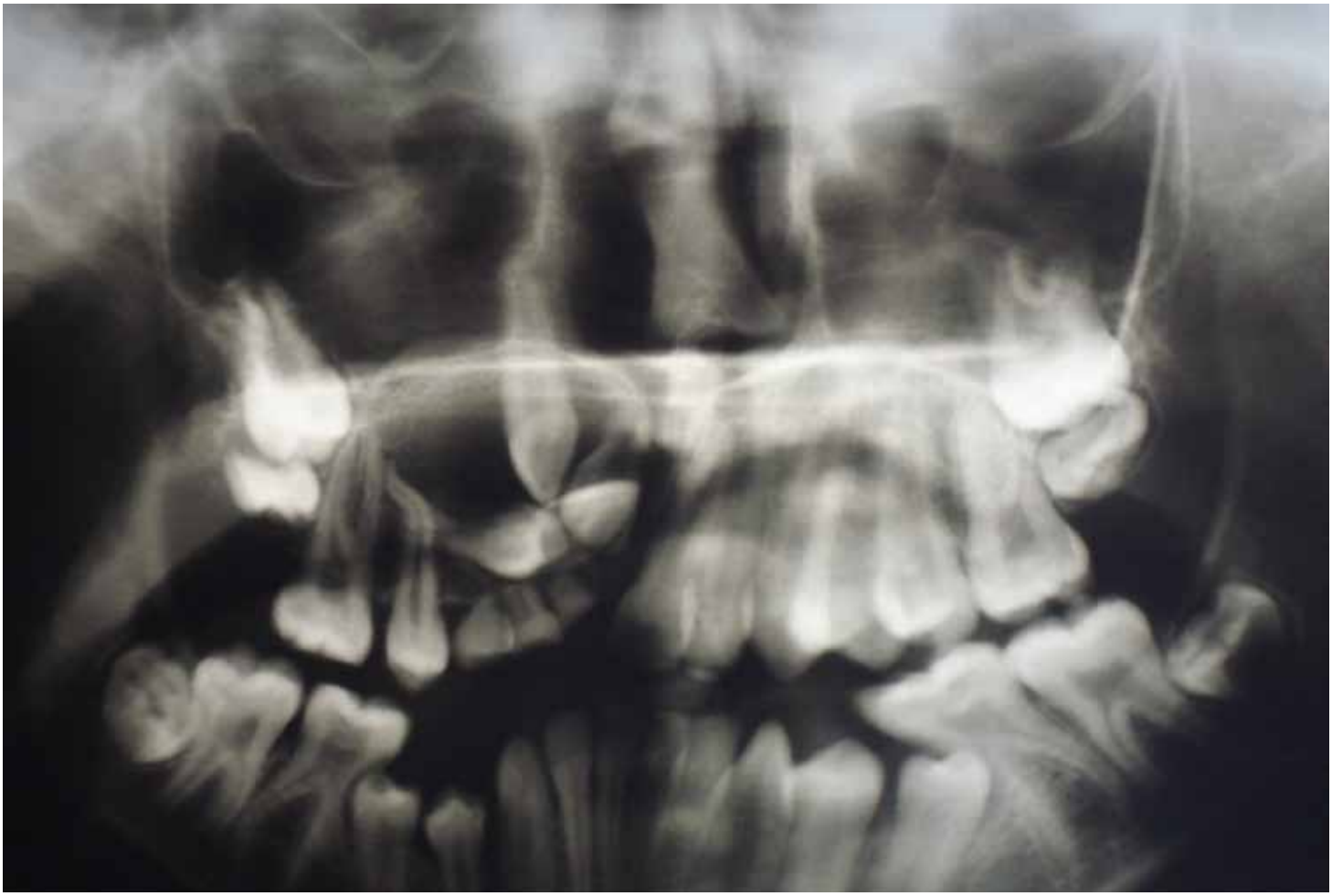
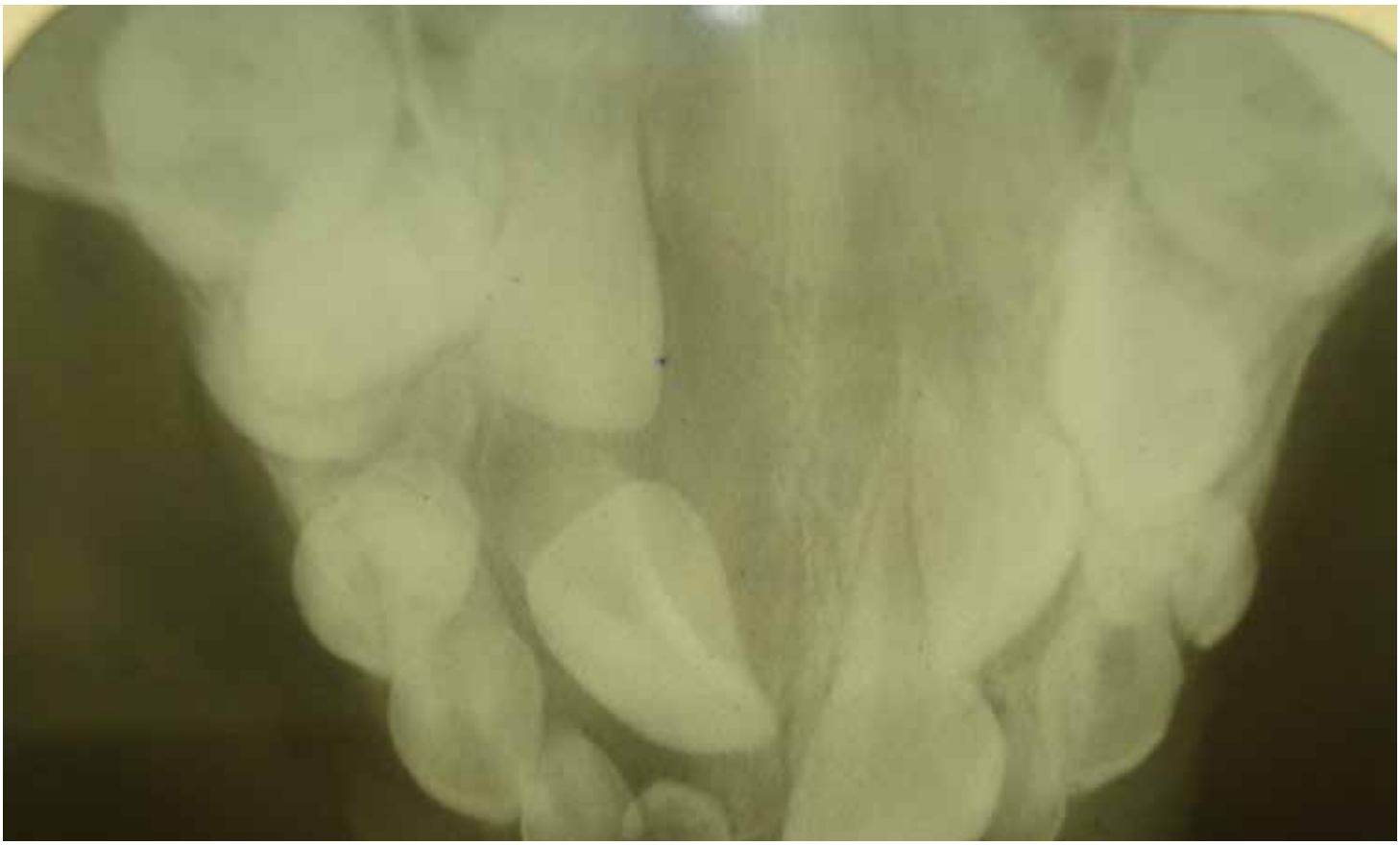


Figure 2. An Upper Standard Occlusal (USO) radiograph showing a retained 52,53 and 54 and the palatally displaced 11,12,13 and 14.



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